Short/branched chain acyl-CoA dehydrogenase deficiency

Description

Short/branched chain acyl-CoA dehydrogenase (SBCAD) deficiency (also known as 2-methylbutyryl-CoA dehydrogenase deficiency) is a rare disorder in which the body is unable to process proteins properly. Normally, the body breaks down proteins from food into smaller parts called amino acids. Amino acids can be further processed to provide energy for the body. People with SBCAD deficiency cannot process a particular amino acid called isoleucine.

Most cases of SBCAD deficiency are detected shortly after birth by newborn screening, which identifies abnormal levels of certain compounds in the blood. In individuals with this condition, a compound called 2-methylbutyryl carnitine is elevated in the blood and another called 2-methylbutyrylglycine is elevated in the urine (2-methylbutyrylglycinuria).

Most people with SBCAD deficiency have no health problems related to the disorder. A small percentage of affected individuals develop signs and symptoms of the condition, which can begin soon after birth or later in childhood. The initial symptoms often include poor feeding, lack of energy (lethargy), vomiting, and irritability. These symptoms sometimes progress to serious health problems such as difficulty breathing, seizures, and coma. Additional problems can include poor growth, vision impairment, learning disabilities, muscle weakness, and delays in motor skills such as standing and walking.

It is unclear why some people with SBCAD deficiency develop health problems and others do not. Doctors suggest that in some cases, signs and symptoms may be triggered by infections, prolonged periods without food (fasting), or an increased amount of protein-rich foods in the diet.

Frequency

SBCAD deficiency is a rare condition; its worldwide prevalence is unknown. This condition is most common among Hmong populations in Southeast Asia and in people of Hmong descent, affecting 1 in 250 to 1 in 500 people in these communities. These individuals do not usually develop health problems related to the condition.

Causes

Mutations in the *ACADSB* gene cause SBCAD deficiency. This gene provides instructions for making an enzyme called short/branched chain acyl-CoA

dehydrogenase (SBCAD), which performs a chemical reaction that helps process the amino acid isoleucine. Mutations in the *ACADSB* gene reduce or eliminate the activity of this enzyme. With a shortage (deficiency) of SBCAD activity, the body is unable to break down isoleucine properly. Researchers speculate that some features of this disorder, such as lethargy and muscle weakness, occur because isoleucine is not converted to energy. In addition, impairment of SBCAD may allow the buildup of toxic compounds, which can lead to serious health problems.

Learn more about the gene associated with Short/branched chain acyl-CoA dehydrogenase deficiency

- ACADSB

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- 2-MBADD
- 2-MBCD deficiency
- 2-MBG
- 2-methylbutyryl glycinuria
- 2-methylbutyryl-CoA dehydrogenase deficiency
- 2-methylbutyryl-coenzyme A dehydrogenase deficiency
- SBCADD
- Short/branched-chain acyl-CoA dehydrogenase deficiency

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Deficiency of 2-methylbutyryl-CoA dehydrogenase (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1864912/)

Genetic and Rare Diseases Information Center

- 2-methylbutyryl-CoA dehydrogenase deficiency (https://rarediseases.info.nih.gov/diseases/10322/2-methylbutyryl-coa-dehydrogenase-deficiency)

Patient Support and Advocacy Resources

- Disease InfoSearch (https://www.diseaseinfosearch.org/)
- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- 2-METHYLBUTYRYL-CoA DEHYDROGENASE DEFICIENCY (https://omim.org/entry/610006)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%282-methylbutyryl-coenzyme+A+dehydrogenase+deficiency%29+OR+%282-methylbutyrylglycine%5BTIAB%5D%29+OR+%28sbcadd%5BTIAB%5D%29+OR+%28short/branched-chain+acyl-coa+dehydrogenase+deficiency%29+OR+%282-MBG%5BTIAB%5D%29+OR+%282-MBCDase%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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